Moyamoya disease

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A 60-year-old previously healthy woman was brought to the emergency department with new-onset coma. Plain computed tomography of the head showed intraventricular hemorrhage and hydrocephalus (Appendix 1, www.cmaj.ca/lookup/suppl/doi:10.1503/cmaj.180681/-/DC1). Extraventricular drainage restored consciousness. Six-vessel cerebral angiography showed stenosis of the circle of Willis, with abundant hazy collateral networks, suggesting moyamoya vasculopathy (Figure 1; video available in Appendix 2, www.cmaj.ca/lookup/suppl/doi:10.1503/cmaj.180681/-/DC1). There was no underlying condition associated with moyamoya vasculopathy. Despite prompt ventricular drainage, the patient was left with severe cognitive impairment. She had no stroke recurrence during a 12-month follow-up period.

Moyamoya vasculopathy is a progressive cerebrovascular disease characterized by stenosis of the circle of Willis with compensatory development of hazy collaterals that resemble a puff of smoke, called "moyamoya" in Japanese. Isolated cases are called moyamoya disease, whereas cases with underlying arteriosclerosis, infectious disease, hematologic disorder, autoimmune disease, neurocutaneous syndrome, chromosomal disorder, cranial irradiation or brain tumour are called moyamoya syndrome.[1] Moyamoya predominantly occurs in East Asian populations, and the *RNF213* gene is considered an important susceptibility factor.[2] The incidence is 0.94 per million population in Japan, but 0.086 per million in the United States.[3,4] The incidence peaks in two age groups: five- to nine-year-old children and adults in their 40s, with a female-to-male ratio of about two, and familial occurrence.[3,4] Although common presentations include transient ischemic attack, ischemic stroke, hemorrhage and headache, moyamoya rarely presents with seizure, choreiform movement, visual deficit, syncope, cognitive impairment or psychiatric symptoms.[1] In children, ischemic events may be triggered by crying with hyperventilation.[1] There is no definitive treatment to reverse or stabilize the primary disease process. Surgical revascularization has been shown to be effective in preventing ischemic stroke, and may have a role in preventing hemorrhagic stroke.[5]

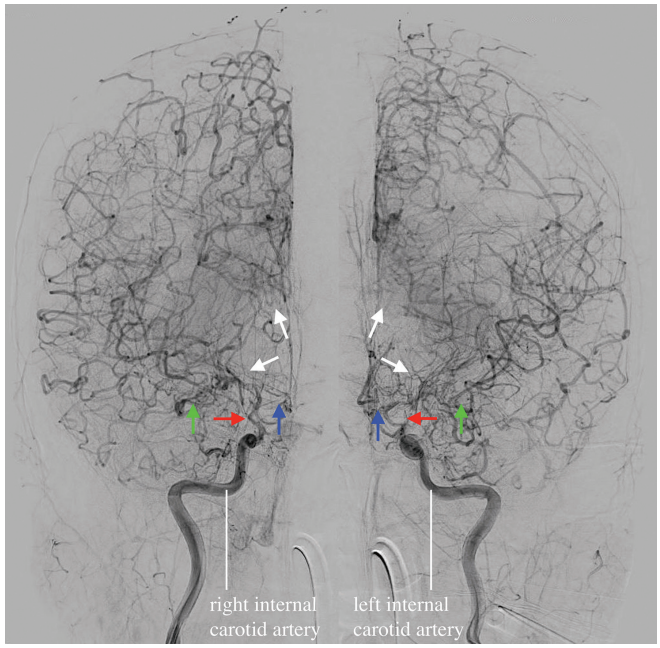

Figure 1: Internal carotid angiography in a 60-year-old woman with acute stroke showing stenosis of the distal internal carotid (red arrows), the anterior cerebral (blue arrows), and the middle cerebral (green arrows) arteries on both sides, with abundant hazy collateral networks (white arrows).

References

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Please see the following video online: Moyamoya vasculopathy in a patient with new-onset coma. www.cmaj.ca/lookup/suppl/doi:10.1503/cmaj.180681/-/DC1

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The author has obtained consent from the patient's family.

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